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E3 56 --> PERLIN M/AU
E4 2 PERLIN M D/AU
E5 52 PERLIN M H/AU
E6 2 PERLIN M J/AU
E7 19 PERLIN M L/AU
E8 1 PERLIN M P/AU
E9 6 PERLIN M S/AU
E10 27 PERLIN M W/AU
E11 5 PERLIN MARK/AU
E12 11 PERLIN MARK W/AU

=> e
E13 11 PERLIN MICHAEL H/AU
E14 6 PERLIN MICHAEL L/AU
E15 1 PERLIN P/AU
E16 1 PERLIN R/AU
E17 5 PERLIN R R/AU
E18 24 PERLIN S/AU
E19 8 PERLIN S A/AU
E20 8 PERLIN S I/AU
E21 4 PERLIN S J/AU
E22 5 PERLIN S M/AU
E23 2 PERLIN SUSAN A/AU
E24 2 PERLIN T/AU

=> s e3, e10, e11, e12
L1 99 ("PERLIN M"/AU OR "PERLIN M W"/AU OR "PERLIN MARK"/AU OR "PERLIN
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L2 2 L1 AND MIXED

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PROCESSING COMPLETED FOR L2
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L3 ANSWER 1 OF 1 MEDLINE DUPLICATE 1
AN 2001668388 MEDLINE
DN 21571107 PubMed ID: 11714147
TI Linear mixture analysis: a mathematical approach to resolving
mixed DNA samples.

AU **Perlin M W; Szabady B**
 CS Cybergenetics, Pittsburgh, PA 15213, USA.
 SO JOURNAL OF FORENSIC SCIENCES, (2001 Nov) 46 (6) 1372-8.
 Journal code: 0375370. ISSN: 0022-1198.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS Priority Journals
 EM 200112
 ED Entered STN: 20011121
 Last Updated on STN: 20020123
 Entered Medline: 20011207
 AB With the advent of PCR-based STR typing systems, **mixed** samples can be separated into their individual DNA profiles. Quantitative peak information can help in this analysis. However, despite such advances, forensic mixture analysis still remains a laborious art, with the high cost and effort often precluding timely reporting. We introduce here a new automated approach to resolving forensic DNA mixtures. Our linear mixture analysis (LMA) is a straightforward mathematical approach that can integrate all the quantitative PCR data into a single rapid computation. LMA has application to diverse mixture problems. As demonstrated here on laboratory STR data, LMA can assess the quality and utility of its solutions. Such rapid and robust methods for computer-based analysis of DNA mixtures may help in reducing crime.

=> s l1 and (pcr or rflp or repeat)
 L4 21 L1 AND (PCR OR RFLP OR REPEAT)
 => s l1 and (pcr or rflp or snp or repeat)
 L5 21 L1 AND (PCR OR RFLP OR SNP OR REPEAT)

=> duplicate remove l5
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 PROCESSING COMPLETED FOR L5
 L6 14 DUPLICATE REMOVE L5 (7 DUPLICATES REMOVED)

=> d 1-14 bib ab

L6 ANSWER 1 OF 14 MEDLINE DUPLICATE 1
 AN 2002199470 IN-PROCESS
 DN 21929801 PubMed ID: 11933190
 TI Determining sequence length or content in zero, one, and two dimensions.
 AU **Perlin Mark W; Szabady Beata**
 CS Cybergenetics, Pittsburgh, Pennsylvania.
 SO HUMAN MUTATION, (2002 Apr) 19 (4) 361-73.
 Journal code: 9215429. ISSN: 1098-1004.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS IN-PROCESS; NONINDEXED; Priority Journals
 ED Entered STN: 20020405
 Last Updated on STN: 20020405
 AB High-throughput assays are essential for the practical application of mutation detection in medicine and research. Moreover, such assays should produce informative data of high quality that have a low-error rate and a low cost. Unfortunately, this is not currently the case. Instead, we typically witness legions of people reviewing imperfect data at astronomical expense yielding uncertain results. To address this problem, for the past decade we have been developing methods that exploit the inherent quantitative nature of DNA experiments. By generating high-quality data, careful DNA-signal quantification permits robust analysis for determining true alleles and certainty measures. We will explore several assays and methods. In a one-dimensional readout, short

tandem **repeat** (STR) data display interesting artifacts. Even with high-quality data, **PCR** artifacts such as stutter and relative amplification can confound correct or automated scoring. However, by appropriate mathematical analysis, these artifacts can be essentially removed from the data. The result is fully automated data scoring, quality assessment, and new types of DNA analysis. These approaches enable the accurate analysis of pooled DNA samples, for both genetic and forensic applications. On a two-dimensional surface (comprised of zero-dimensional spots) one can perform assays of extremely high-throughput at low cost. The question is how to determine DNA sequence length or content from nonelectrophoretic intensity data. Here again, mathematical analysis of highly quantitative data provides a solution. We will discuss new lab assays that can produce data containing such information; mathematical transformation then determines DNA length or content.

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L6 ANSWER 2 OF 14 MEDLINE DUPLICATE 2
 AN 2001668388 MEDLINE
 DN 21571107 PubMed ID: 11714147
 TI Linear mixture analysis: a mathematical approach to resolving mixed DNA samples.
 AU **Perlin M W**; Szabady B
 CS Cybergenetics, Pittsburgh, PA 15213, USA.
 SO JOURNAL OF FORENSIC SCIENCES, (2001 Nov) 46 (6) 1372-8.
 Journal code: 0375370. ISSN: 0022-1198.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS Priority Journals
 EM 200112
 ED Entered STN: 20011121
 Last Updated on STN: 20020123
 Entered Medline: 20011207
 AB With the advent of **PCR**-based STR typing systems, mixed samples can be separated into their individual DNA profiles. Quantitative peak information can help in this analysis. However, despite such advances, forensic mixture analysis still remains a laborious art, with the high cost and effort often precluding timely reporting. We introduce here a new automated approach to resolving forensic DNA mixtures. Our linear mixture analysis (LMA) is a straightforward mathematical approach that can integrate all the quantitative **PCR** data into a single rapid computation. LMA has application to diverse mixture problems. As demonstrated here on laboratory STR data, LMA can assess the quality and utility of its solutions. Such rapid and robust methods for computer-based analysis of DNA mixtures may help in reducing crime.

L6 ANSWER 3 OF 14 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
 AN 2000:466443 BIOSIS
 DN PREV200000466443
 TI Method and system for genotyping.
 AU **Perlin, Mark W.** (1)
 CS (1) 5904 Beacon St., Pittsburgh, PA, 15217 USA
 PI US 6054268 April 25, 2000
 SO Official Gazette of the United States Patent and Trademark Office Patents, (Apr. 25, 2000) Vol. 1233, No. 4, pp. No pagination. e-file.
 ISSN: 0098-1133.
 DT Patent
 LA English
 AB The present invention pertains to a process which can be fully automated for accurately determining the alleles of genetic markers. More specifically, the present invention is related to performing **PCR** amplification on locations of DNA to generate a reproducible pattern, labeling the **PCR** products, converting the labels into a signal, operating on the signal, and then determining the genotype of the location of the DNA. An amplification can include multiple locations from the DNA

of one or more individuals. The invention also pertains to genetics applications and systems which can effectively use this genotyping information.

L6 ANSWER 4 OF 14 MEDLINE DUPLICATE 3
AN 1999455102 MEDLINE
DN 99455102 PubMed ID: 10523529
TI Using quality measures to facilitate allele calling in high-throughput genotyping.
AU Palsson B; Palsson F; **Perlin M**; Gudbjartsson H; Stefansson K; Gulcher J
CS deCODE Genetics, Inc., 110 Reykjavik, Iceland.
SO GENOME RESEARCH, (1999 Oct) 9 (10) 1002-12.
Journal code: CES; 9518021. ISSN: 1088-9051.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 199912
ED Entered STN: 20000113
Last Updated on STN: 20000113
Entered Medline: 19991202
AB Currently, the main limitation in high-throughput microsatellite genotyping is the required manual editing of allele calls. Even though programs for automated allele calling have been available for several years, they have limited capability because accurate data could only be assured by manual inspection of the electropherograms for confirmation. Here we describe the development of a parametric approach to allele call quality control that eliminates much of the time required for manual editing of the data. This approach was implemented in an editing tool, Decode-GT, that works downstream of the allele calling program, TrueAllele (TA). Decode-GT reads the output data from TA, displays the underlying electropherograms for the genotypes, and sorts the allele calls into three categories: good, bad, and ambiguous. It discards the bad calls, accepts the good calls, and suggests that the user inspect the ambiguous calls, thereby reducing dependence on manual editing. For the categorization we use the following parameters: (1) the quality value for each allele call from TrueAllele; (2) the peak height of the alleles; and (3) the size of the peak shift needed to move peaks into the nearest bin. Here we report how we optimized the parameters such that the size of the ambiguous category was minimized, and both the number of miscalled genotypes in the good category and the useable genotypes in the bad category were negligible. This approach reduces the manual editing time and results in <1% miscalls.

L6 ANSWER 5 OF 14 MEDLINE DUPLICATE 4
AN 96026116 MEDLINE
DN 96026116 PubMed ID: 7485172
TI Toward fully automated genotyping: genotyping microsatellite markers by deconvolution.
AU **Perlin M W**; Lancia G; Ng S K
CS Computer Science Department, Carnegie Mellon University, Pittsburgh, PA 15213, USA.
NC R01 NS32084 (NINDS)
SO AMERICAN JOURNAL OF HUMAN GENETICS, (1995 Nov) 57 (5) 1199-210.
Journal code: 3IM; 0370475. ISSN: 0002-9297.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 199511
ED Entered STN: 19960124
Last Updated on STN: 19960124
Entered Medline: 19951130
AB Dense genetic linkage maps have been constructed for the human and mouse

genomes, with average densities of 2.9 cM and 0.35 cM, respectively. These genetic maps are crucial for mapping both Mendelian and complex traits and are useful in clinical genetic diagnosis. Current maps are largely comprised of abundant, easily assayed, and highly polymorphic PCR-based microsatellite markers, primarily dinucleotide (CA)_n repeats. One key limitation of these length polymorphisms is the PCR stutter (or slippage) artifact that introduces additional stutter bands. With two (or more) closely spaced alleles, the stutter bands overlap, and it is difficult to accurately determine the correct alleles; this stutter phenomenon has all but precluded full automation, since a human must visually inspect the allele data. We describe here novel deconvolution methods for accurate genotyping that mathematically remove PCR stutter artifact from microsatellite markers. These methods overcome the manual interpretation bottleneck and thereby enable full automation of genetic map construction and use. New functionalities, including the pooling of DNAs and the pooling of markers, are described that may greatly reduce the associated experimentation requirements.

L6 ANSWER 6 OF 14 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
 AN 1995:266918 BIOSIS
 DN PREV199598281218
 TI Towards fully automated genotyping: Use of an X linked recessive spastic paraplegia family to test alternative analysis methods.
 AU Kobayashi, H.; Hoffman, E. P. (1); Matise, T. C.; Perlin, M. W.; Marks, H. G.
 CS (1) Dep. Molecular Genetics Biochem., Human Genetics Pediatrics, Univ. Pittsburgh Sch. Med., Pittsburgh, PA 15261 USA
 SO Human Genetics, (1995) Vol. 95, No. 5, pp. 483-490.
 ISSN: 0340-6717.
 DT Article
 LA English
 AB Advances in dinucleotide-based genetic maps open possibilities for large scale genotyping at high resolution. The current rate-limiting steps in use of these dense maps is data interpretation (allele definition), data entry, and statistical calculations. We have recently reported automated allele identification methods. Here we show that a 10-cM framework map of the human X chromosome can be analyzed on two lanes of an automated sequencer per individual (10-12 loci per lane). We use this map and analysis strategy to generate allele data for an X-linked recessive spastic paraplegia family with a known PLP mutation. We analyzed 198 genotypes in a single gel and used the data to test three methods of data analysis: manual meiotic breakpoint mapping, automated concordance analysis, and whole chromosome multipoint linkage analysis. All methods pinpointed the correct location of the gene. We propose that multipoint exclusion mapping may permit valid inflation of LOD scores using the equation max LOD - (next best LOD).

L6 ANSWER 7 OF 14 MEDLINE DUPLICATE 5
 AN 96015064 MEDLINE
 DN 96015064 PubMed ID: 8530043
 TI Rapid construction of integrated maps using inner product mapping: YAC coverage of human chromosome 11.
 AU Perlin M W; Duggan D J; Davis K; Farr J E; Findler R B; Higgins M J; Nowak N J; Evans G A; Qin S; Zhang J; +
 CS Computer Science Department, Carnegie Mellon University, Pittsburgh, Pennsylvania 15213, USA.
 NC HG00856 (NHGRI)
 MH00802 (NIMH)
 SO GENOMICS, (1995 Jul 20) 28 (2) 315-27.
 Journal code: GEN; 8800135. ISSN: 0888-7543.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS Priority Journals
 EM 199602

ED Entered STN: 19960220
 Last Updated on STN: 19980206
 Entered Medline: 19960201

AB Inner product mapping (IPM) has been proposed as a hybridization-based method for achieving low-cost, high-throughput, high-resolution radiation hybrid (RH) mapping of clones. Using Alu-PCR products of chromosome 11-specific clones, we serially hybridized a set of RHs against gridded filters of YACs having an average size of 350 kb. We then combined these hybridization data with preexisting RH map data to build an inner product map. This binning of 865 YACs provides the first high-resolution large-scale (> twofold redundancy) clonal coverage of human chromosome 11 and is the first inner product map ever constructed. We verified the accuracy and precision of this chromosome 11 map by performing a novel likelihood analysis on independent YAC hybridization data. These results establish that IPM is a highly rapid, inexpensive, accurate, and precise large-scale long-range mapping method, particularly when preexisting RH maps are available, and that IPM can replace or complement more conventional short-range mapping methods. IPM may enable the rapid construction of sequence-ready maps and the binning of expressed sequences.

L6 ANSWER 8 OF 14 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
 AN 1995:476445 BIOSIS
 DN PREV199598490745
 TI Quantitative analysis of gel electrophoresis data for automated genotyping applications.
 AU Richards, D. R.; Perlin, M. W.
 CS Computer Sci. Dep., Carnegie Mellon Univ., Pittsburgh, PA USA
 SO American Journal of Human Genetics, (1995) Vol. 57, No. 4 SUPPL., pp. A26. Meeting Info.: 45th Annual Meeting of the American Society of Human Genetics Minneapolis, Minnesota, USA October 24-28, 1995
 ISSN: 0002-9297.
 DT Conference
 LA English

L6 ANSWER 9 OF 14 MEDLINE DUPLICATE 6
 AN 95029378 MEDLINE
 DN 95029378 PubMed ID: 7942856
 TI Toward fully automated genotyping: allele assignment, pedigree construction, phase determination, and recombination detection in Duchenne muscular dystrophy.
 AU Perlin M W; Burks M B; Hoop R C; Hoffman E P
 CS School of Computer Science, Carnegie Mellon University, Pittsburgh, PA 15213.
 NC R01 NS32084 (NINDS)
 SO AMERICAN JOURNAL OF HUMAN GENETICS, (1994 Oct) 55 (4) 777-87.
 Journal code: 3IM; 0370475. ISSN: 0002-9297.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS Priority Journals
 EM 199410
 ED Entered STN: 19941222
 Last Updated on STN: 20000303
 Entered Medline: 19941025

AB Human genetic maps have made quantum leaps in the past few years, because of the characterization of > 2,000 CA dinucleotide **repeat** loci: these PCR-based markers offer extraordinarily high PIC, and within the next year their density is expected to reach intervals of a few centimorgans per marker. These new genetic maps open new avenues for disease gene research, including large-scale genotyping for both simple and complex disease loci. However, the allele patterns of many dinucleotide **repeat** loci can be complex and difficult to interpret, with genotyping errors a recognized problem. Furthermore, the possibility of genotyping individuals at hundreds or thousands of

polymorphic loci requires improvements in data handling and analysis. The automation of genotyping and analysis of computer-derived haplotypes would remove many of the barriers preventing optimal use of dense and informative dinucleotide genetic maps. Toward this end, we have automated the allele identification, genotyping, phase determinations, and inheritance consistency checks generated by four CA repeats within the 2.5-Mbp, 10-cM X-linked dystrophin gene, using fluorescein-labeled multiplexed PCR products analyzed on automated sequencers. The described algorithms can deconvolute and resolve closely spaced alleles, despite interfering stutter noise; set phase in females; propagate the phase through the family; and identify recombination events. We show the implementation of these algorithms for the completely automated interpretation of allele data and risk assessment for five Duchenne/Becker muscular dystrophy families. The described approach can be scaled up to perform genome-based analyses with hundreds or thousands of CA-repeat loci, using multiple fluorophors on automated sequencers.

L6 ANSWER 10 OF 14 MEDLINE
 AN 96039037 MEDLINE
 DN 96039037 PubMed ID: 7584409
 TI Intelligent DNA-based molecular diagnostics using linked genetic markers.
 AU Pathak D K; Hoffman E P; Perlin M W
 CS Department of Computer Science, Carnegie Mellon University, USA.
 SO ISMB, (1994) 2 331-9.
 Journal code: CCP; 9509125.
 CY United States
 DT Journal; Article; (JOURNAL ARTICLE)
 LA English
 FS Priority Journals
 EM 199512
 ED Entered STN: 19960124
 Last Updated on STN: 19960124
 Entered Medline: 19951214
 AB This paper describes a knowledge-based system for molecular diagnostics, and its application to fully automated diagnosis of X-linked genetic disorders. Molecular diagnostic information is used in clinical practice for determining genetic risks, such as carrier determination and prenatal diagnosis. Initially, blood samples are obtained from related individuals, and PCR amplification is performed. Linkage-based molecular diagnosis then entails three data analysis steps. First, for every individual, the alleles (i.e., DNA composition) are determined at specified chromosomal locations. Second, the flow of genetic material among the individuals is established. Third, the probability that a given individual is either a carrier of the disease or affected by the disease is determined. The current practice is to perform each of these three steps manually, which is costly, time consuming, labor-intensive, and error-prone. As such, the knowledge-intensive data analysis and interpretation supersede the actual experimentation effort as the major bottleneck in molecular diagnostics. By examining the human problem solving for the task, we have designed and implemented a prototype knowledge-based system capable of fully automating linkage-based molecular diagnostics in X-linked genetic disorders, including Duchenne Muscular Dystrophy (DMD). Our system uses knowledge-based interpretation of gel electrophoresis images to determine individual DNA marker labels, a constraint satisfaction search for consistent genetic flow among individuals, and a blackboard-style problem solver for risk assessment. We describe the system's successful diagnosis of DMD carrier and affected individuals from raw clinical data.

L6 ANSWER 11 OF 14 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
 AN 1994:525359 BIOSIS
 DN PREV199497538359
 TI Automated genotyping of dinucleotide repeat markers.
 AU Perlin, M. W. (1); Hoffman, E. P.

CS (1) Computer Sci., Carnegie Mellon Univ., Pittsburgh, PA USA
SO American Journal of Human Genetics, (1994) Vol. 55, No. 3 SUPPL., pp. A199.
Meeting Info.: 44th Annual Meeting of the American Society of Human Genetics Montreal, Quebec, Canada October 18-22, 1994
ISSN: 0002-9297.

DT Conference
LA English

L6 ANSWER 12 OF 14 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 1993:444537 BIOSIS
DN PREV199345080162
TI Detection of indicator bacteria and pathogens in water by polymerase chain reaction (PCR) and gene probe methods.
AU Atlas, R. M. (1); Bej, A. (1); Mahbubani, M. (1); Steffan, R. (1); Perlin, M. (1); Dicesare, J.; Haff, L.
CS (1) Dep. Biol., Univ. Louisville, Louisville, KY USA
SO Mongkolsuk, S. [Editor]; Lovett, P. S. [Editor]; Trempey, J. E. [Editor]. (1992) pp. 219-223. Biotechnology and environmental science: Molecular approaches.
Publisher: Plenum Press 233 Spring Street, New York, New York, USA.
Meeting Info.: International Conference Bangkok, Thailand August 21-24, 1990
ISBN: 0-306-44352-X.

DT Article
LA English

L6 ANSWER 13 OF 14 MEDLINE DUPLICATE 7
AN 92152848 MEDLINE
DN 92152848 PubMed ID: 1785923
TI Detection of Giardia cysts by using the polymerase chain reaction and distinguishing live from dead cysts.
AU Mahbubani M H; Bej A K; Perlin M; Schaefer F W 3rd; Jakubowski W; Atlas R M
CS Department of Biology, University of Louisville, Kentucky 40292.
SO APPLIED AND ENVIRONMENTAL MICROBIOLOGY, (1991 Dec) 57 (12) 3456-61.
Journal code: 6K6; 7605801. ISSN: 0099-2240.
CY United States
DT Journal; Article; (JOURNAL ARTICLE)
LA English
FS Priority Journals
EM 199203
ED Entered STN: 19920405
Last Updated on STN: 19920405
Entered Medline: 19920316

AB A method was developed for the detection of Giardia cysts by using the polymerase chain reaction (PCR) and the giardin gene as the target. DNA amplification by PCR, using giardin DNA as the target, resulted in detection of both live and dead cysts. When giardin mRNA was used as the target, the ability to amplify cDNA by PCR depended on the mode of killing. Cysts killed by freezing were not detected by PCR when giardin mRNA was the target. Cysts killed by heating or exposure to monochloramine, however, gave positive detection signals for both DNA and giardin mRNA targets. The amount of giardin mRNA and total RNA was significantly increased in live cysts following the induction of excystation. Cysts killed by freezing, heating, or exposure to monochloramine did not show a change in RNA content. The detection of the giardin gene by PCR permits a sensitive and specific diagnosis for Giardia spp. Discrimination between live and dead cysts can be made by measuring the amounts of RNA or PCR-amplified product from the giardin mRNA target before and after the induction of excystation.

L6 ANSWER 14 OF 14 BIOSIS COPYRIGHT 2002 BIOLOGICAL ABSTRACTS INC.
AN 1991:381182 BIOSIS

DN BR41:53572
TI DETECTION OF VIABLE LEGIONELLA AND GIARDIA USING PCR AND GENE
PROBES.
AU MAHBUBANI M; BEJ A; PERLIN M; SCHAEFER F; ATLAS R M
CS UNIV. LOUISVILLE, LOUISVILLE, KY.
SO 91ST GENERAL MEETING OF THE AMERICAN SOCIETY FOR MICROBIOLOGY 1991,
DALLAS, TEXAS, USA, MAY 5-9, 1991. ABSTR GEN MEET AM SOC MICROBIOL. (1991)
91 (0), 87.
CODEN: AGMME8.
DT Conference
FS BR; OLD
LA English

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COST IN U.S. DOLLARS	SINCE FILE ENTRY	TOTAL SESSION
FULL ESTIMATED COST	17.46	17.67

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